Overview of the Genetic Disease Branch Newborn Screening Program

The Newborn Screening Program begins with the education of prenatal care providers and hospital staff. The prenatal care providers distribute a copy of the informational booklet, *Important Information for Parents about the Newborn Screening Test*, to women during their pregnancy. This booklet describes mandatory newborn screening. Because some women do not receive prenatal care, the booklet is also provided to women upon admission to a licensed perinatal health facility for delivery. The actual sample of newborn blood is obtained after 12 hours of age and before the sixth day of life by a heel-stick. The blood sample is collected on special filter paper, and mailed to the pre-assigned regional state contract laboratory for testing. These laboratories process the specimen and enter demographic data and test results on terminals linked to a Genetic Disease Branch central computer in Richmond, California. A computer-generated written report of all tests, referred to as a "result mailer" is mailed to the hospital that collected the specimen. Another copy is mailed to the baby's physician as listed by the hospital of birth on the Newborn Screening Specimen Collection Form (also referred to as the "test request form" or TRF).

Because of the urgency for treatment, all initial positive test results for PKU, galactosemia and primary congenital hypothyroidism are immediately reported by telephone by the NAPS lab to assigned newborn screening coordinators. These coordinators are located at one of the seven state-funded Area Service Centers (ASC) and are linked to the state central computer in Richmond, CA. The State Genetic Disease Laboratory (GDL) reviews and releases hemoglobin results. Potentially clinically significant hemoglobinopathies and other initial positive results are reported on a daily "Interesting Case Report" to the Centers. The coordinators from the Centers immediately telephone the newborn's physician to provide interpretation of the test and explain necessary follow-up. Medical consultants and specialists are also available to provide additional information and consultation when necessary.
If the initial tests are not within normal limits, a second blood sample called a "recall specimen" is collected (see attached flow chart). The recall specimens for phenylketonuria are sent to GDL for testing. Recall specimens for galactosemia are sent to a state-contract galactosemia confirmation laboratory at Children's Hospital of Los Angeles, while those for hemoglobin disorders are sent to the state hemoglobin reference laboratory at Children's Hospital and Research Center at Oakland. In the past, filter paper recall specimens for TSH were also sent to GDL for testing. As of February 2005, the state laboratory will no longer conduct recall testing on filter paper blood specimens for primary congenital hypothyroidism, and providers will be required to have a venous blood specimen collected/tested at a private California licensed laboratory (including hospital laboratories)**.** Test results will then be reported to the NBS Program via the assigned (ASC).

The NBS coordinators notify the newborn's physician of the recall test results. Because primary care physicians are often unfamiliar with these rare disorders, the coordinator will assist the provider in referring a family to a California Children's Services (CCS)-approved metabolic, endocrine or sickle cell disease

center/hematology center for specialized diagnosis and treatment. The comprehensive team approach to care offered at the Centers is particularly important in treatment of these disorders.

The coordinators also directly contact the parents of the newborn by telephone to discuss the need for confirmatory testing or a diagnostic evaluation. After contacting the parents, the coordinator faxes a referral form to the appropriate CCS office requesting an authorization for diagnostic evaluation at a CCS approved center, or in certain circumstances, a CCS paneled specialist (e.g. pediatric endocrine, pediatric hematologist) not affiliated with a center. The coordinator schedules the appointment at the center and will send the parents a letter to the mother containing information about CCS and CCS application form, with instructions for submission and the date of the appointment. It is anticipated that this process will expedite the referral and the care of the newborn. Once care is authorized, the center will make the arrangements for any further confirmatory/diagnostic testing and develop if needed the treatment plan.